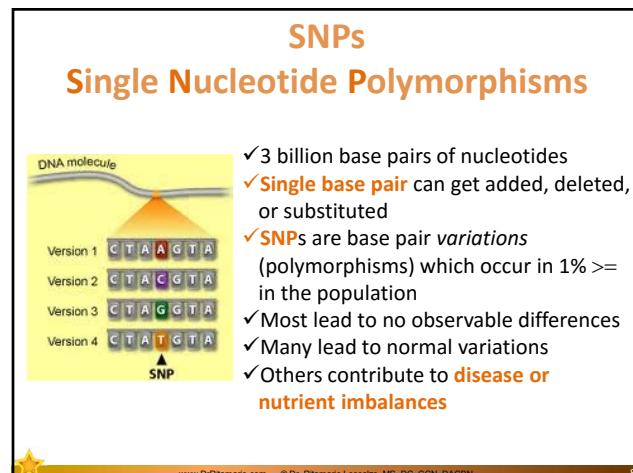
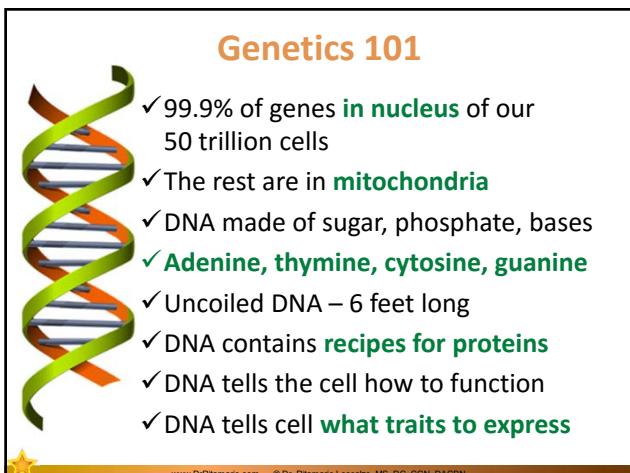
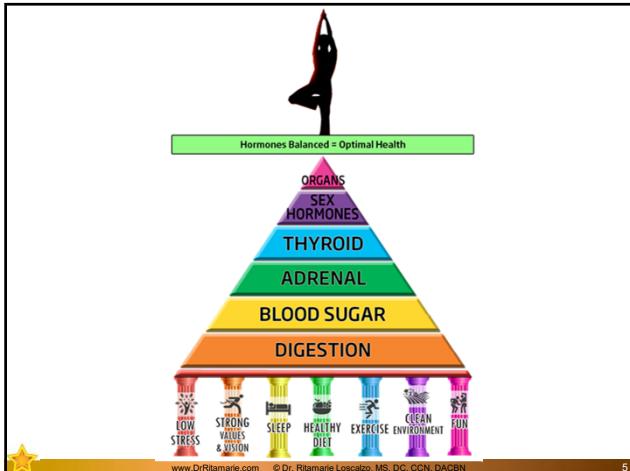




**Medical Disclaimer:** The information in this presentation is not intended to replace a one-on-one relationship with a qualified health care professional and is not intended as medical advice. It is intended as a sharing of knowledge and information from the research and experience of Dr. Ritamarie Loscalzo, drritamarie.com, and the experts who have contributed. We encourage you to make your own health care decisions based upon your research and in partnership with a qualified health care professional.



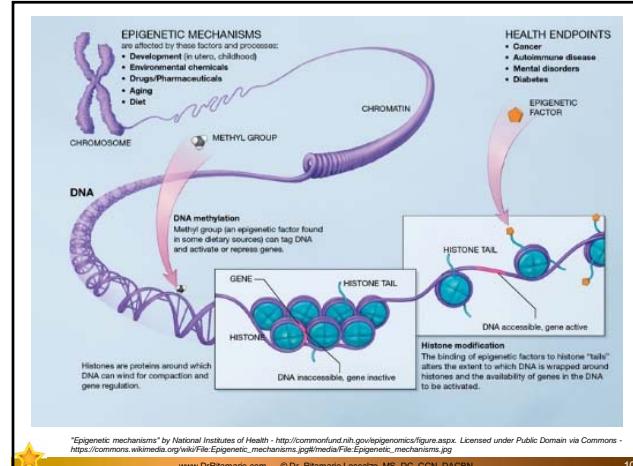


## Epigenetics



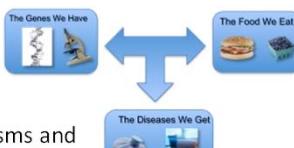
- ✓ Heritable changes in gene expression that does not involve changes to the underlying DNA sequence
- ✓ A change in phenotype without a change in genotype
- ✓ Modulated by histones and methyl groups

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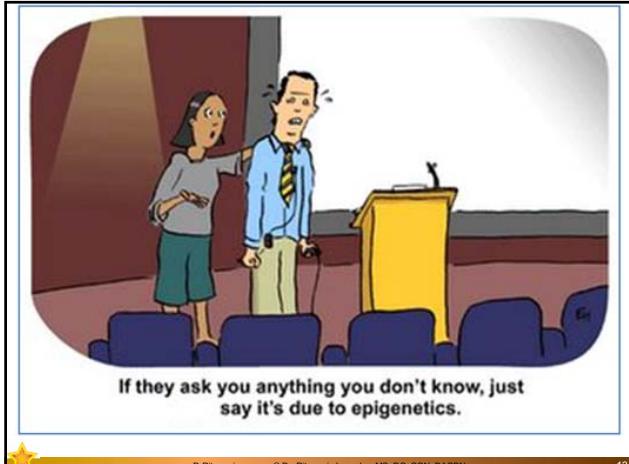
## Nutrigenomics

- ✓ Study of the **effects of foods and nutrients on gene expression**
- ✓ The influence of genetic variation on nutrition
- ✓ The correlation between single-nucleotide polymorphisms and a **nutrient's absorption, metabolism, elimination or biological effects**
- ✓ Experts say 5% of genetic expression is truly tied to our genes; the **remaining 95% is within your control**




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13

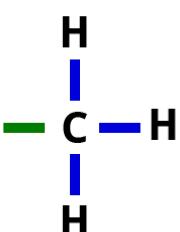
## Methylation Functions

- ✓ Turn on and off genes
- ✓ Process **chemicals, endogenous toxins, and xenobiotic compounds**
- ✓ Build neurotransmitters (norepinephrine, epinephrine, serotonin, melatonin)
- ✓ **Metabolize neurotransmitters** (dopamine, epinephrine)
- ✓ Process hormones (estrogen)
- ✓ Build immune cells (T cells, NK cells)
- ✓ **Synthesize DNA bases**
- ✓ Produce energy (CoQ10, carnitine, creatine, ATP)
- ✓ Produce protective coating on nerves (**myelination**)
- ✓ Build and maintain **cell membranes** (phosphatidylcholine)
- ✓ Repair damaged DNA



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## Methylation

  
**Methyl group**

**Methyl Donors**

- SAM-e
- Folate
- Vitamin B12
- TMG
- DMG

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## Main Methylation SNPs

- ✓ MTHFR C677T
- ✓ MTHFR 1298C
- ✓ MTRR, MTR
- ✓ BHMT
- ✓ CBS
- ✓ COMT



Approximately 45% of the population has 1 copy of the MTHFR C677T SNP

Approximately 90% of those with chronic disease have 1 copy of the MTHFR C677T

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## Gut-Brain Conditions Associated With Methylation SNPs



- ✓ Fibromyalgia
- ✓ Chronic fatigue syndrome
- ✓ Autism
- ✓ Depression
- ✓ Insomnia
- ✓ ADD/ADHD
- ✓ Irritable bowel syndrome
- ✓ Inflammatory bowel syndrome
- ✓ Migraine
- ✓ Raynaud's
- ✓ Cancer
- ✓ Alzheimer's
- ✓ Parkinson's

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## SNPs Related to the Gut-Brain Axis

SNP	Possible impairments
TAS2R38	Eating disinhibition
ANKK1/DRD2	Effects on amount of effort put out to obtain food
TAS2R38	Bitter taster gene
TAS1R3	Sweet tooth gene
FTO	Effect on not feeling satisfied after eating
LEPR	Leptin receptor gene – associated with snacking behavior
SLC2A2	Sweet tooth
NBPF3	Risk of vitamin B6 deficiency – cofactor for neurotransmitter synthesis
SLC23A1	Risk of decreased vitamin C and increased risk of gastric cancer and IBD
MAO-A	Catalyzes deamination of dopamine, norepinephrine, and serotonin. Associated with a variety of psychiatric disorders, including antisocial behavior, obsessive compulsive disorders and anxiety.
GAD	Involved in the conversion of glutamate to GABA
LRRK2	Linked to increased risk of Parkinson's
IgA	Immune protection for GI and other mucous membranes

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## SNPs Related to the Gut-Brain Axis

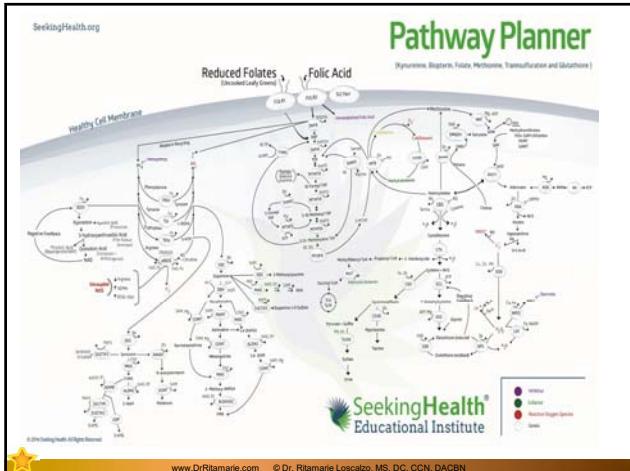
SNP	Description
MC4R	Regulates energy balance
COMT	Can cause IBS due to too much dopamine
CYP27B1	Encodes an enzyme that activates vitamin D
DHCR7	Related to cholesterol and vitamin D synthesis
MTHFR	Affects folate metabolism, which is important for gut repair and methylation
HLA DQ2	Celiac disease and gluten sensitivity risk – most common gene as 90% of all celiac patients have it
HLA DQ8	Celiac disease risk – less prevalent than DQ2
ATG16L1	Increased risk of Crohn's disease
ApoA4	Role in lipid absorption in the intestines.
FUT2	Impact on the ability to secrete ABO antigens in body fluids, i.e., saliva, sweat, tears, gut. "Non-secretors" need more bifidobacteria as they can't make the oligosaccharide that feeds them. On the positive side, non-secretors are more resistant to H. pylori and rotavirus and have a higher risk for Crohn's.
DAO	Histamine breakdown
ACE	Regulates fluid balance and blood pressure

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## SNPs Related to the Gut-Brain Axis

SNP	Description
CYP2E1	Related to alcohol metabolism and effects on liver and pancreas
APOA2	Regulates after meal response to saturated fat. C variant – saturated fats WILL make you fat, T variant + saturated fat - WILL NOT make you fat
GSTM3 V224I	Association with late-onset Alzheimer's disease
APOE	Association with Alzheimer's disease and ability to process saturated fat
CYP2C19	Increased risk of GERD
CCL26	Reflux; Eosinophilic esophagitis
ADRB3	Possible link to increased risk of gallstones and gallbladder cancer
PPARGC1A	Associated with non-alcoholic fatty liver disease
MCM6	Associated with lactose intolerance
GC	Encodes an enzyme that transports vitamin D in blood to cells. Risk of decreased vitamin D, which affects composition of the bacterial flora in the gut microbiome
VDR	Vitamin D receptor gene
FADS1	Risk of decreased omega-3 and omega-6 fatty acids

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## Before Addressing Individual SNPs

### ✓ Foundational lifestyle/diet

- Hydration
- Clean, antioxidant-rich, whole foods diet
- Reduce stress

### ✓ Gut healing and pathogen removal

### ✓ Address mitochondrial dysfunction

### ✓ Remove all folic acid

- ✓ B vitamin support
- ✓ Balance blood sugar
- ✓ Adaptogens and adrenal support
- ✓ Test homocysteine levels
- ✓ Thyroid support



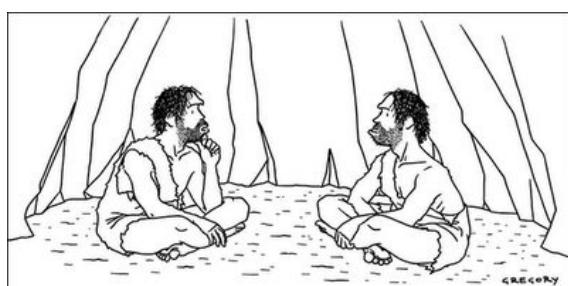
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## Genomic Testing



- ❑ 23 and Me
  - ✓ <http://www.23andme.com>
- ❑ Genova Diagnostics
  - ✓ <http://www.GenovaDiagnostics.com>
- ❑ Holistic Health International (Dr. Amy Yasko)
  - ✓ <http://www.HolisticHealth.com>
- ❑ Pathway FIT
  - ✓ <http://www.pathwayfit.com>
- ❑ SpectraCell (MTHFR Only)
  - ✓ <http://www.drritamarie.com/go/SCGenotyping>

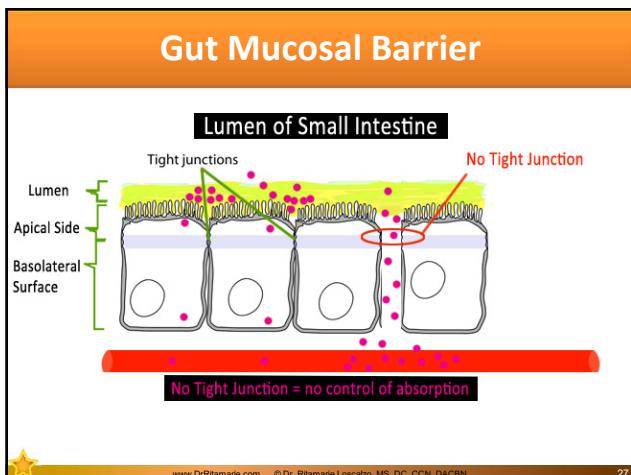
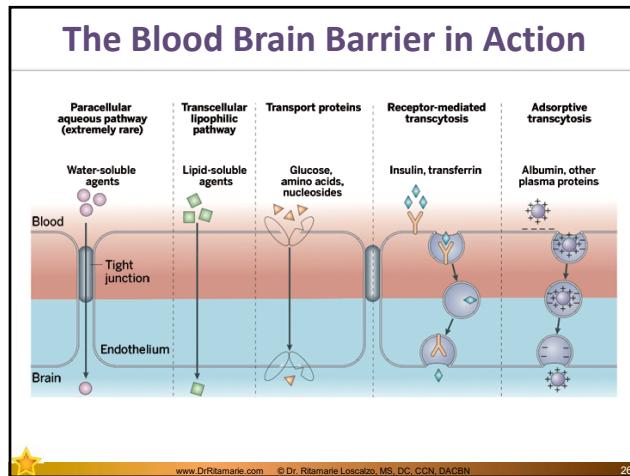
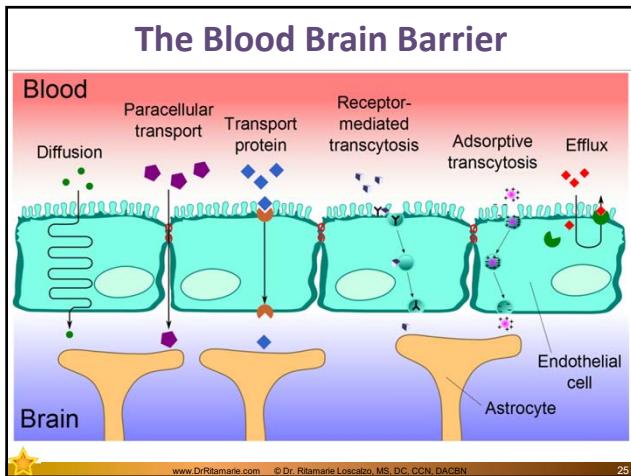
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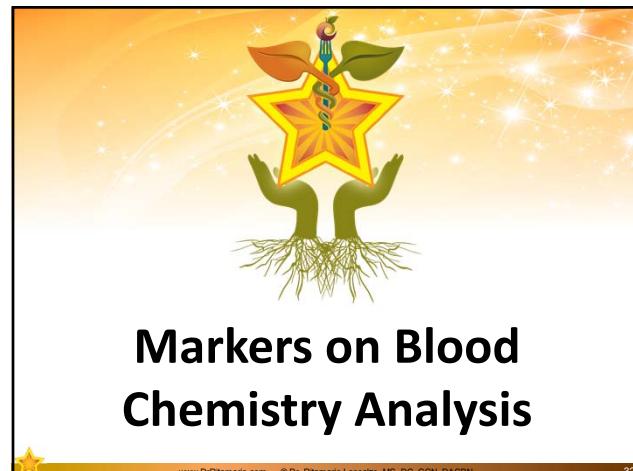
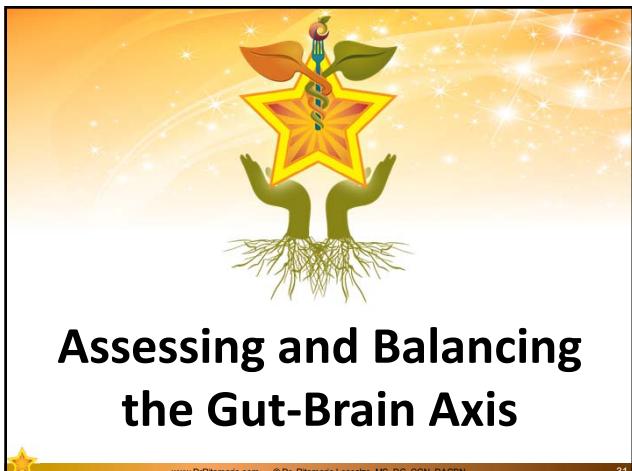
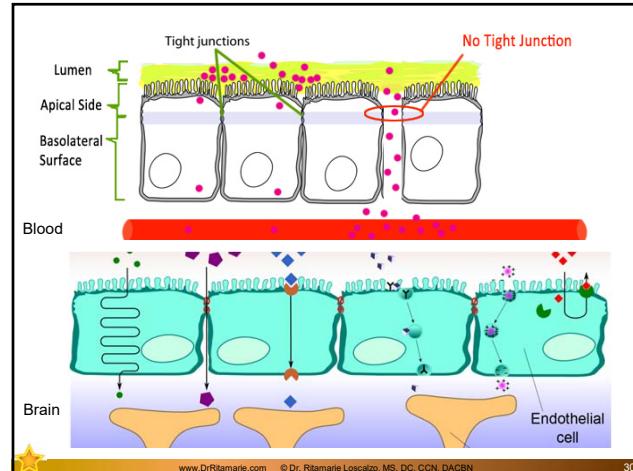
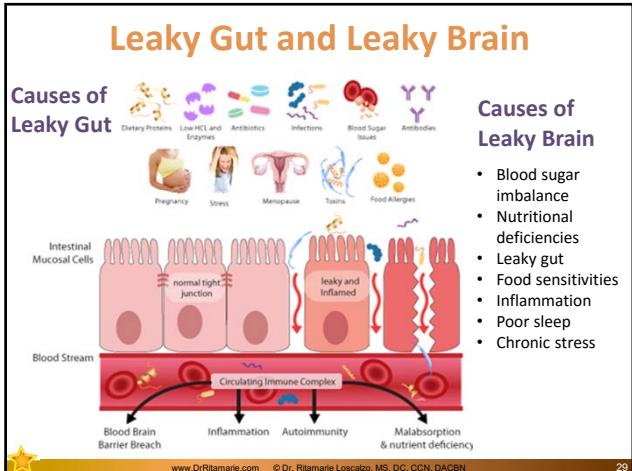


"Something's just not right—our air is clean, our water is pure, we all get plenty of exercise, everything we eat is organic and free-range, and yet nobody lives past thirty."

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24





## Low Stomach Acid

- ✓ BUN (hi or lo)
- ✓ Chloride (lo)
- ✓ Carbon Dioxide (hi)
- ✓ Calcium (lo)
- ✓ Phosphorus (lo)
- ✓ Protein (lo)
- ✓ Albumin (lo)
- ✓ Globulin (hi)
- ✓ Iron (lo)
- ✓ Hemoglobin (lo)
- ✓ MCV (hi)
- ✓ MCH (hi)
- ✓ MCHC (hi)
- ✓ Uric Acid (lo)
- ✓ Ferritin (lo)



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## Stomach

### Hypochlorhydria

- Total Protein (hi or lo)
- Globulin (hi or lo)
- BUN (hi or lo)
- Phosphorus (lo)
- Creatinine (lo)
- Iron (lo)
- Calcium (lo)

**PLUS**

### Helicobacter Pylori

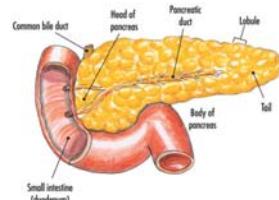
- WBC (hi or lo)
- Neutrophil (hi)
- Monocytes (normal to hi)
- Lymphocytes (lo)
- Special testing for H. pylori and Urea Breath Test



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## Small Intestine and Pancreas

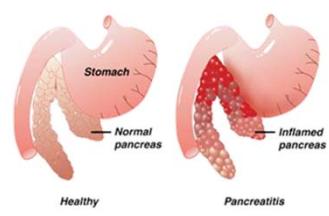
- ✓ Uric Acid (hi)
- ✓ BUN (lo)
- ✓ Creatinine (lo)
- ✓ Protein (lo)
- ✓ Globulin (lo): GI inflammation
- ✓ Alkaline phosphatase (lo)
- ✓ Alkaline Phosphatase (hi): leaky gut
- ✓ GGT (lo): malabsorption
- ✓ Hematocrit (lo): inflammation



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## Pancreatitis

- ✓ Glucose (hi)
- ✓ Triglycerides (hi)
- ✓ Alkaline Phosphatase (hi)
- ✓ Lipase (hi)
- ✓ Amylase (hi)
- ✓ GGTP (hi)
- ✓ SGOT (normal to hi)
- ✓ SGPT (normal to hi)
- ✓ Albumin (lo)



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### Large Intestine

- ✓ Eosinophils (hi) - parasites
- ✓ Sodium (lo)
  - possible laxative use or adrenal issues

**For in-depth look, need to do:**

- ✓ Comprehensive stool and digestive analysis
- ✓ Parasitology
- ✓ Organic Acids Test (OAT) - markers for dysbiosis



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### Home and In-Office Testing

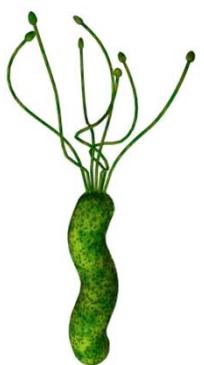
- ✓ Assessment questionnaires and scorecards
- ✓ Physical signs
- ✓ HCl challenge
- ✓ Transit time test



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### H. pylori Symptoms

- ✓ Gnawing or burning abdominal pain, just beneath the ribs; worse on an empty stomach
- ✓ Loss of appetite
- ✓ Bloating, burping
- ✓ Nausea, vomiting
- ✓ Black, tarry stools
- ✓ Autoimmune
- ✓ Cardiovascular, skin, liver, and biliary symptoms



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### Evaluating Gastroesophageal Reflux (GERD, AKA Heartburn)

**Symptoms**

- ✓ Food feels trapped behind breastbone
- ✓ Burning pain in the chest: worse with bending, lying down, at night
- ✓ Nausea after eating
- ✓ Difficulty swallowing
- ✓ Hiccups
- ✓ Hoarseness or change in voice
- ✓ Regurgitation of food
- ✓ Sore throat after eating
- ✓ Possibly: Cough or wheezing

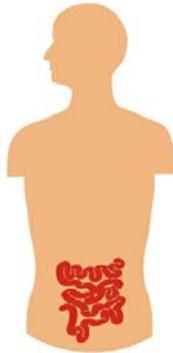
**Possible Causes**

- ✓ Overeating
- ✓ Vigorous exercise after eating
- ✓ Alcohol
- ✓ Caffeine – coffee, chocolate
- ✓ Tight garments
- ✓ Eating while or right before reclining
- ✓ Smoking
- ✓ Stress

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### Small Intestine Physical Signs

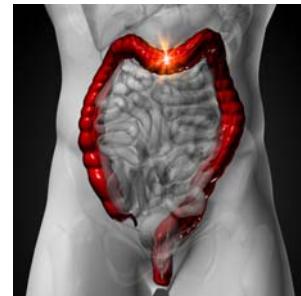
- ✓ Tender spot at tips of 8<sup>th</sup> through 10<sup>th</sup> ribs
- ✓ Tender spot between 7<sup>th</sup> and 8<sup>th</sup> ribs on left
- ✓ Tenderness, hardness, or distension in a square, 3-inch radius in all directions from umbilicus



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### Large Intestine Physical Signs

- ✓ Tenderness along 8<sup>th</sup> through 10<sup>th</sup> ribs
- ✓ Tenderness over large or small intestine
- ✓ Hydration status: hands at side palpate veins
- ✓ Tenderness along outside of thigh



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### Intestinal Function Testing

- ✓ Gluten and casein antibodies – Enterolab, Cyrex Labs
- ✓ Breath test
- ✓ Low zinc on zinc assay test
- ✓ Malabsorption: Urine Indican – Apex Energetics
- ✓ Bowel transit time
- ✓ Pulse test for food reactions, food allergy test
- ✓ Intestinal permeability – Cyrex Labs
- ✓ Bowel transit time
- ✓ Stool testing for dysbiosis, gut function – BioHealth, Doctor's Data, Genova Diagnostics, DiagnosTechs



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### HCl Challenge

- ✓ Home test to determine need for stomach acid supplementation.
- ✓ Start with ONE 500-650 mg capsule (not tablet) containing both hydrochloric acid (HCl) and 150 mg of pepsin.
- ✓ Take HCl after a few bites of food; do not take on an empty stomach or after meals.
- ✓ If you have no discomfort (burning or warm sensation), add one capsule per meal.
- ✓ If you experience pain, burning, or a warm sensation, take one of the following:
  - Take 1 teaspoon slippery elm in 8 ounces warm water
  - ¼ cup aloe vera juice
  - ¼ teaspoon baking soda in water
- ✓ Next meal, go back to the dose that caused no pain.

**DO NOT** go above the maximal dose of 80 grains for the average man and 60 grains for the average sized woman.



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### Bowel Transit Time Tracking

- ✓ Swallow 4 charcoal capsules at evening meal. Record the date and time
- ✓ After each bowel movement, observe your stool for the first sign of black or grey. Observe in a brightly lit room.
- ✓ Calculate the number of hours between "Time Charcoal Taken" to "Time/Date Color First Appears".
- ✓ Continue to observe every stool and note the time and date when the color has completely disappeared. Ideally, it should take between 18 and 24 hours.
- ✓ Wait 5 days to make sure all is completely disappeared and try again.
- ✓ Repeat with a variety of test meals.



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45

### Comprehensive Digestive Analysis and Dysbiosis Testing

- ✓ Stool samples
- ✓ Multiple days
- ✓ Labs
  - BioHealth
  - Doctor's Data
  - DiagnosTechs
  - Genova Diagnostics
- ✓ Ubiome



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46

### Assessing Leaky Gut and Leaky Brain

- **Occulin/Zonulin:** signaling protein that transiently and reversibly opens the tight junctions between intestinal cells.
- **ActoMyosin:** a protein that anchors the tight junctions and provides structure for the intestinal muscle cells.
- **Lipopolysaccharides (LPS):** endotoxin produced by gram-negative bacteria upon death which can damage the intestinal lining and lead to leaky gut.
- **Blood Brain Barrier Protein:** tests for integrity of blood brain barrier.



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47

### Organic Acids Test - Great Plains

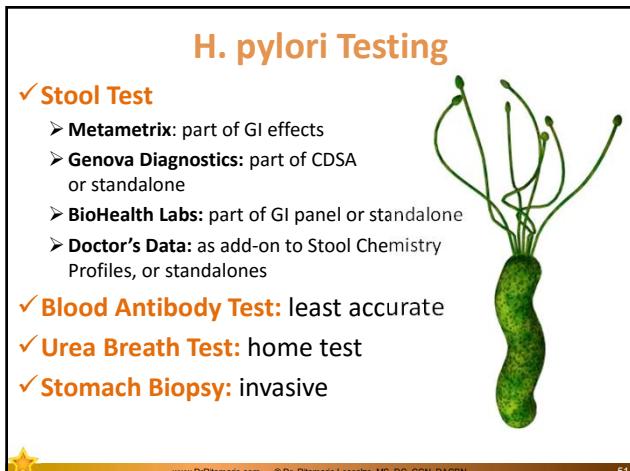
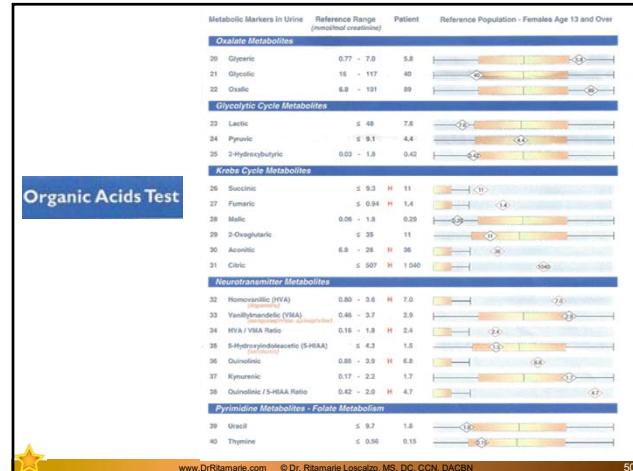
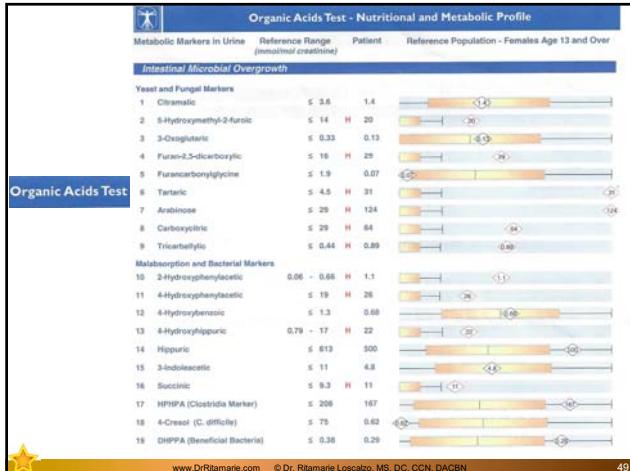
- ✓ Intestinal Microbial Overgrowth
- ✓ Oxalate Metabolites
- ✓ Glycolytic Metabolites
- ✓ Kreb Cycle Metabolites
- ✓ Neurotransmitter Metabolites
- ✓ Folate Metabolism
- ✓ Ketones and Fatty Acid Oxidation
- ✓ Nutritional Markers
- ✓ Detoxification Indicators
- ✓ Amino Acid Metabolites
- ✓ Bone Metabolites

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48



### Case: 40-year-old Female

- ✓ **History:** 3-year-old and a nursing infant, running a successful online business
- ✓ **Top Presenting Complaints:** Fatigue, headaches, pelvic pain, poor sleep, inability to lose weight, brain fog, and "mommy brain"
- ✓ **Medications:** Naturthroid, Advil on occasion
- ✓ **Supplements:** Lots

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53

### Case: 40-year-old Female

- ✓ **Diet:**
  - Mostly whole foods, organic
  - Regularly: Raw cheese, sprouted grain bread (gluten), grains, eggs, chocolate, sweets, decaf coffee, green smoothies, greens, and salads a few days a week; no alcohol
  - Had done a raw vegan diet and felt great and lost weight, but felt starved all the time
- ✓ **Exercise:** Sporadic due to exhaustion and overwhelm with infant and toddler

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54

### Case: 40-year-old Female

- ✓ **Prior Labs:** TSH optimal, TG 151, HDL 36, TPO 14, candida blood antibody screen negative
- ✓ **Initial Testing:** Low vitamin D (22), low adrenal function, low thyroid function and Free T4 1, Free T3 2.2 (poor T4 to T3 conversion), gluten intolerance
- ✓ **Suspected:** Insulin resistance, leptin resistance, gluten intolerance, multiple food allergies, leaky gut, adrenal fatigue
- ✓ **Approach:** Started with foundations, gluten-free, dairy-free, and monitoring blood sugar
- ✓ **Advanced Testing:** Details follow

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55

### Case: 40-year-old Female

- ✓ **Genetics Testing:**
  - MTHFR C677T: homozygous
  - MTR: homozygous
  - MTRR: heterozygous
  - BHMT: homozygous
  - ACHY: heterozygous
  - COMT: heterozygous
  - MAO: homozygous
  - CBS: homozygous

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56

## Enterolab: Stool Gluten Sensitivity

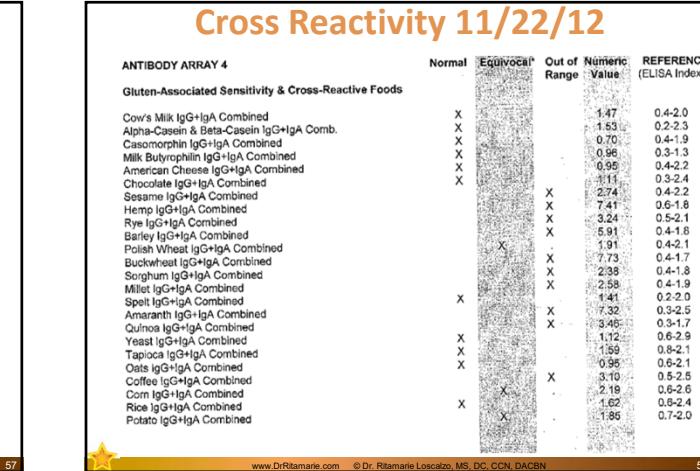
### Gluten Sensitivity Stool Test

Fecal Anti-gliadin IgA 13 Units (Normal Range is less than 10 Units)

**Interpretation of Fecal Anti-gliadin IgA:** The level of intestinal anti-gliadin IgA antibody was elevated, indicative of active dietary gluten sensitivity. For optimal health: resolution or improvement of gluten-induced syndromes (mainly falling into six categories abbreviated as NAAAGS – neuropsychiatric, autoimmune, asthma, abdominal, glandular deficiencies/hyperactivity or skin diseases); resolution of symptoms known to be associated with gluten sensitivity (such as abdominal symptoms - pain, cramping, bloating, gas, diarrhea and/or constipation, chronic headaches, chronic sinus congestion, depression, arthritis, chronic skin problems/rashes, fibromyalgia, and/or chronic fatigue); and prevention of small intestinal damage and malnutrition, osteoporosis, and damage to other tissues (like nerves, brain, joints, muscles, thyroid, pancreas, other glands, skin, liver, spleen, among others), it is recommended that you follow a strict and permanent gluten free diet. As gluten sensitivity is a genetic syndrome, you may want to have your relatives screened as well.

**Later confirmed by Alcat testing and elimination/provocation**

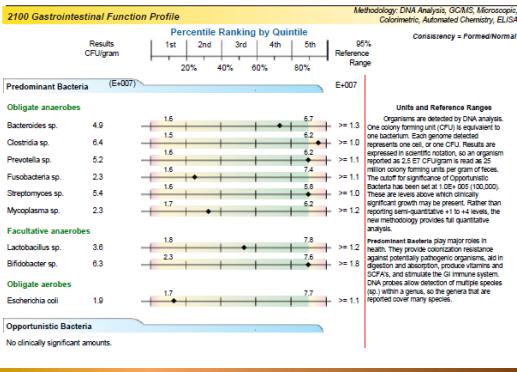
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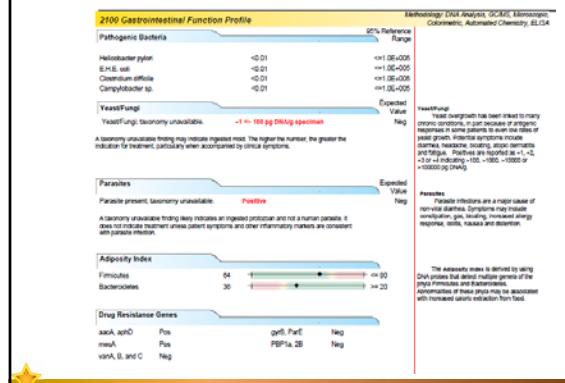
58

## Metametrix/Genova Test 12/9/11



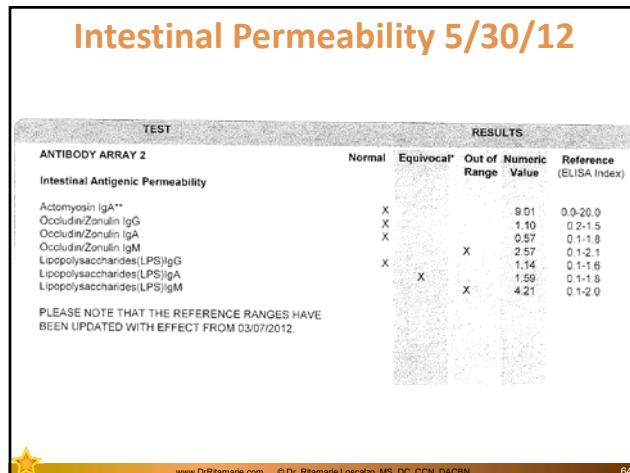
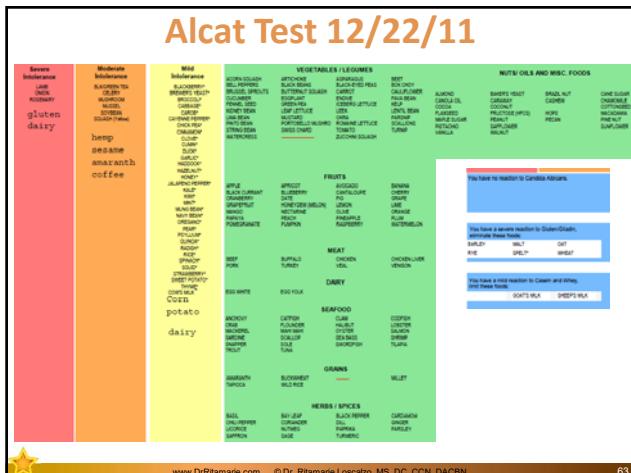
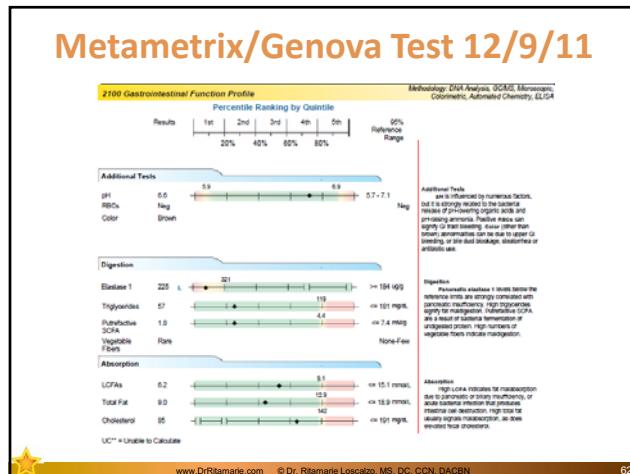
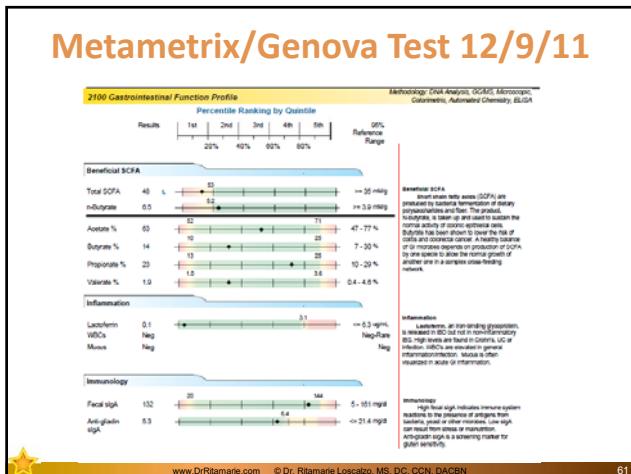
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## Metametrix/Genova Test 12/9/11



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60



**Initial 6/12/12**

## GI Pathogen Screen with H. pylori Antigen - 401H

Parameter	Result
<b>***Stool Culture for Pathogens***</b>	
Source	Stool
Preliminary Report	Normal flora after 24 hours
Final Report	* Klebsiella species isolated *
Amount of Growth	Moderate
<b>***Stool for Ova &amp; Parasites***</b>	
Ova & parasites #1	No Ova/Parasites detected
Ova & parasites #2	No Ova/Parasites detected
Ova & parasites #3	* Endolimax nana cysts detected *
Ova & parasites #4	No Ova/Parasites detected
Trichrome Stain	No Ova/Parasites detected
<b>***Stool Antigens Test***</b>	
Cryptosporidium Antigen	Not detected
E. histolytica Antigen	Not detected
Giardia lamblia Antigen	Not detected
...	
Fungi	No fungi isolated
C. difficile Toxin A	Not detected
C. difficile Toxin B	Not detected
Yeast	No yeasts isolated
Occult Blood	Not detected
<b>***Helicobacter Pylori Stool Antigen***</b>	
H. pylori Antigen	* Detected *

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65

**Follow-Up 8/7/12**

## GI Pathogen Screen with H. pylori Antigen - 401H

Parameter	Result
<b>***Stool Culture for Pathogens***</b>	
Source	Stool
Preliminary Report	Normal flora after 24 hours
Final Report	* Escherichia coli isolated *
Amount of Growth	Abundant
<b>***Stool for Ova &amp; Parasites***</b>	
Ova & parasites #1	* Endolimax nana cysts detected *
Ova & parasites #2	No Ova/Parasites detected
Ova & parasites #3	No Ova/Parasites detected
Ova & parasites #4	* Endolimax nana cysts detected *
Trichrome Stain	Few cyst forms of Endolimax nana seen on Trichrome Stain
<b>***Stool Antigens Test***</b>	
Cryptosporidium Antigen	Not detected
E. histolytica Antigen	Not detected
Giardia lamblia Antigen	Not detected
...	
Fungi	No fungi isolated
C. difficile Toxin A	Not detected
C. difficile Toxin B	Not detected
Yeast	No yeasts isolated
Occult Blood	Not detected
<b>***Helicobacter Pylori Stool Antigen***</b>	
H. pylori Antigen	Not detected

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66

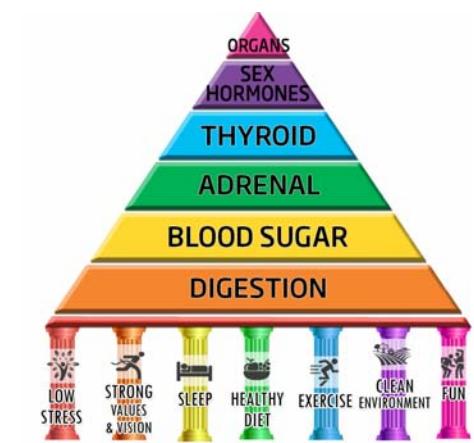
**Follow-Up 10/8/12**

## GI Pathogen Screen with H. pylori Antigen - 401H

Parameter	Result
<b>***Stool Culture for Pathogens***</b>	
Source	Stool
Preliminary Report	Normal flora after 24 hours
Final Report	* Escherichia coli isolated *
Amount of Growth	Abundant
<b>***Stool for Ova &amp; Parasites***</b>	
Ova & parasites #1	No Ova/Parasites detected
Ova & parasites #2	No Ova/Parasites detected
Ova & parasites #3	No Ova/Parasites detected
Ova & parasites #4	No Ova/Parasites detected
Trichrome Stain	No Ova/Parasites detected
<b>***Stool Antigens Test***</b>	
Cryptosporidium Antigen	Not detected
E. histolytica Antigen	Not detected
Giardia lamblia Antigen	Not detected
...	
Fungi	No fungi isolated
C. difficile Toxin A	Not detected
C. difficile Toxin B	Not detected
Yeast	No yeasts isolated
Occult Blood	Not detected
<b>***Helicobacter Pylori Stool Antigen***</b>	
H. pylori Antigen	Not detected

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67



## SEX HORMONES

## THYROID

## ADRENAL

## BLOOD SUGAR

## DIGESTION

## LOW

## STRESS

## STRONG

## VALUES

## &amp;

## VISION

## SLEEP

## HEALTHY

## DIET

## EXERCISE

## ENVIRONMENT

## CLEAN

## FUN